J Ped Surg Case Reports 2 (2014) 274–276

Contents lists available at ScienceDirect

Journal of Pediatric Surgery CASE REPORTS

journal homepage: www.jpscasereports.com

Poor prognosis myofibromatosis in a 3 year old male☆

Xinghai Yang*, Shigang Cheng, Hongqiong Geng, Yifan Zhang

Department of Pediatric Surgery, Hubei Women and Children's Hospital, Wuhan 430070, PR China

ARTICLE INFO

Article history:
Received 27 April 2014
Received in revised form
18 May 2014
Accepted 20 May 2014

Key words:
Myofibromatosis
Fibrous proliferation
Nodules

ABSTRACT

Infantile myofibromatosis (IM) is a very rare clinical entity consisting of fibrous proliferation occurring in infants and children. We report the case of a male child with the occurrence of multiple soft tissue nodules. Myofibromatosis was diagnosed on histopathological examination of the lesions. The characteristic clinical, radiologic and histopathologic features of this process are reviewed along with diagnostic and therapeutic options.

IM is a rare tumor of infancy and childhood, typically presenting as a firm, nodular mass involving soft tissue, bone or viscera [1,2]. Approximately one-third of cases involve the head and neck. IM occurs in solitary, multiple, and generalized forms, with similar histology but different clinicopathologic and prognostic implications. We describe the case of a boy with myofibromatosis presenting with multiple nodules in the skin, muscle, and subcutaneous tissues of the trunk.

1. Case report

A three year-old boy presented with a mass on the left side of neck, which had been expanding over the previous month.

The child was born at full term gestation with a birth weight of 3150 g, and was initially noted to have similar migrating masses on the forehead and occipital regions in the first month of life. Sometimes these masses would disappear spontaneously. The exact diagnosis eluded several other hospitals.

The family history was unremarkable. He underwent a right laparoscopic orchidopexy at the age of two and half years, had never traveled, and was otherwise healthy. He had no history of injury with good health and normal growth.

The child was found to have a firm mobile 4 cm mass on the left side of his neck in other hospital a month ago. There was no erythema nor other skin changes. He remained afebrile. The mass continued to slowly expand despite applying local physiotherapy on the local lesion and intravenous antibiotics in another hospital.

1.1. Physical examination

Priot to his presentation and through his evaluation he had normal vital signs. He had a soft tissue nodule extending from the left shoulder to the occiput about 12 × 7 cm in size, which was approximately 2 cm raised above the surface of skin. The quality of the mass was moderately firm, without redness, with local swelling and tenderness. There was no fluctuance nor or pitting with finger pressure. The boundaries were indistinct. Blood tests to assess complete blood count (CBC), lever functions, and renal function were all normal. Paragonimiasis skin test was negative. Cervical vertebra and chest x-ray, as well as ultrasound evaluation of liver, gallbladder, and kidneys were unremarkable. CT scan was concerning for inflammation causing the soft tissue swelling on the back of his neck.

Because of rapid enlargement of the mass, a full thickness biopsy was performed on the lesion including overlying skin, fascia and degenerative muscle for pathologic diagnosis. Histological examination showed that there was proliferation of capillaries, lymphatic vessels and fat tissue, which suggested inflammation and seemed to exclude the possibility of malignant tumor. He was treated with anti-inflammatories after the biopsy. A new painless mass rapidly appeared on his back and grew to 20 × 6.2 cm in 5 days, while the original neck lesion began to diminish. The patient underwent partial surgical removal of the newly appeared lesion. Histological

* Corresponding author. Tel.: +86 27 87169259.
E-mail address: xinghai_yang@hotmail.com (X. Yang).

2213-5766/$ – see front matter © 2014 The Authors. Published by Elsevier Inc. All rights reserved.
http://dx.doi.org/10.1016/j.epsc.2014.05.010

and biochemical examination demonstrated histopathological features of IM (Fig. 1). The spindle cells showed a myogenic character with strongly positive alpha-smooth muscle actin staining.

After the second biopsy, a progressive increase occurred in the lesions which involved the right neck and the left anterior of chest wall. Antibiotics, anti-inflammatories and traditional Chinese medicine were not successful in slowing the growth of the lesions.

Seven years in out-patient follow-up revealed that the lesions continued to grow and new lesions appeared with multiple nodules on the back, all lesions were firm and elevated (Fig. 2). Both sides of the shoulder joint and neck were involved with limitation of motion.

2. Discussion

Infantile myofibromatosis was first reported by Stout as congenital generalized fibromatosis in 1954 [3]. Several names were used for describing IM, including the following: multiple mesenchymal hamartoma, multiple vascular leiomyoma of the newborn, diffuse congenital fibromatosis, congenital multiple fibromatosis, generalized hamartomatosis, and multiple congenital mesenchymal tumor. The term infantile myofibromatosis was first coined by Chun and Enzinger in 1981 [4].

IM is a descriptive term for a rare benign soft tissue tumor. The etiology of IM is uncertain. Autosomal dominant, recessive and polygenic modes of inheritance have been reported [5,6]. Literature on IM is scant. IM is a mesenchymal disorder presenting multiple soft tissue nodules on the scalp, trunk and subcutaneous tissue, and can affect internal organs such as the liver, kidney and bone

Fig. 2. A boy with multiple nodules in the back was presented.

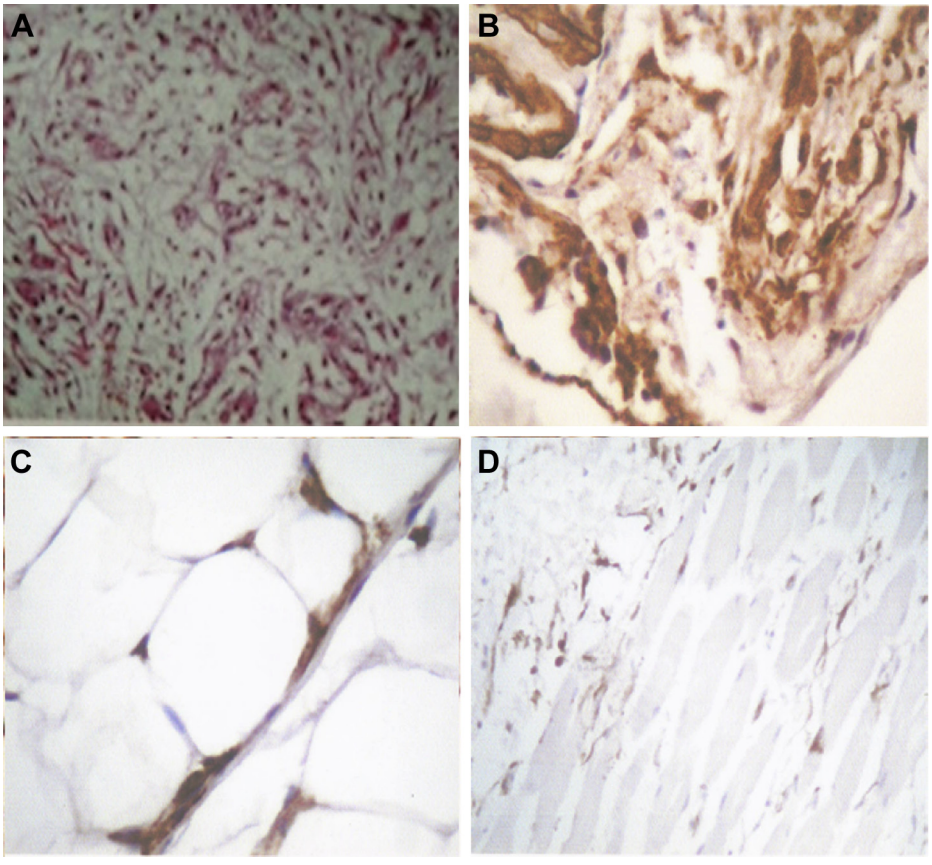

Fig. 1. (A) Proliferation of spindle-shaped or oval fibroblastic cells and fibrous collagen matrix, atrophy of muscle cells (20×). (B) In some areas, spindle cells were rich in blood vessels, which were positive for alpha-smooth muscle actin (20×). (C–D) spindle cells among adipose cells (C) or striated muscle cells (D), which were positive for alpha-smooth muscle actin (C 40×, D 20×).

marrow. IM can lead to vital organ dysfunction and can become life-threatening eventually.

Histologically it is characterized by two cell types arranged in a biphasic pattern [7]; namely centrally positioned small rounded cells with pale staining nuclei and eosinophilic cytoplasm and peripherally elongated spindle cells. The lesions show the features of proliferation of spindle-shaped or oval fibroblastic cells and fibrous collagen matrix, atrophy of muscle cells with hydropic changes, and mucoid or degeneration of collagen matrix in soft tissues. Immunohistochemically the nodules may be found to be positive for alpha smooth muscle actin, but negative for S-100 actin.

Prior reports have documented spontaneous regression of IM. Careful follow-up must be done, as recurrences after regression may occur [8]. Chung reported a 7% recurrence rate [5]. Recurrences after 8 and 15 years have been reported.

No effective treatment has been identified [9]. Antibiotics have been ineffective. In aggressive cases, limited success has been achieved by treatment with radiotherapy, steroid injection, chemotherapy and combined chemo-radiation therapy [10]. Alpha interferon and the anti-estrogen agent tamoxifen have also been used in some patients with infantile myofibromatosis [11,12], but no improvement was seen. There is no consensus between chemotherapy, radiotherapy, steroid injection, interferon therapy.

In our patient, the lesions have slowly involved the shoulder joints and neck of the patient, but is not life threatening at this point. The prognosis may be worse if involvement occurs of visceral organs.

3. Conclusion

IM usually develops at birth or during the first years of life. The presence of alpha smooth muscle actin in the spindle cells indicates the diagnosis of infantile myofibromatosis. The case described here is multicentric IM with multiple nodules in the skin, muscle, and subcutaneous tissues of the trunk.

Conflict of interest statement

The authors have no conflict of interest to disclose.

References

[1] Holzer-Fruehwald L, Blaser S, Rossi A, Fruehwald-Pallamar J, Thurnher MM. Imaging findings in seven cases of congenital infantile myofibromatosis with cerebral, spinal, or head and neck involvement. Neuroradiology 2012;54:1389–98.
[2] Kim EJ, Wang KC, Lee JY, Phi JH, Park SH, Cheon JE, et al. Congenital solitary infantile myofibromatosis involving the spinal cord. J Neurosurg Pediatr 2013;11:82–6.
[3] Stout AP. Juvenile fibromatosis. Cancer 1954;7:953–78.
[4] Chung EB, Enzinger FM. Infantile myofibromatosis. Cancer 1981;48:1807–18.
[5] Bracko M, Cindro L, Golouh R. Familial occurrence of infantile myofibromatosis. Cancer 1992;69:1294–9.
[6] Venencie PY, Bigel P, Desgruelles C, Lortat-Jacob S, Dufier JL, Saurat JH. Infantile myofibromatosis. Report of two cases in one family. Br J Dermatol 1987;117:255–9.
[7] Oliver RJ, Coulthard P, Carre C, Sloan P. Removal of an intraparenchymal infantile myofibroma. Oral Oncol 2003;39:626–9.
[8] Miwa T, Oi S, Nonaka Y, Tamogami R, Sasaki H, Akiyama M, et al. Rapid spontaneous regression of multicentric infantile myofibromatosis in the posterior fossa and lumbar vertebra. Childs Nerv Syst 2011;27:491–6.
[9] Levine E, Fréneaux P, Schleiermacher G, Brisse H, Pannier S, Teissier N, et al. Risk-adaptedtherapy for infantile myofibromatosis in children. Pediatr Blood Cancer 2012;59:115–20.
[10] Gandhi MM, Natha PC, Weitzman S, Levitt GA. Successful treatment of life threatening generalized infantile myofibromatosis using loe-dose chemotherapy. J Pediatr Hematol Oncol 2003;25:750–4.
[11] Savaşan S, Fulgenzi LA, Rabah R, Mohamed AN, Ravindranath Y. Generalized infantile myofibromatosis in a patient with Turner's syndrome: a trial of interferon-alpha. J Pediatr Surg 1998;133:694–6.
[12] Ebert Jr CS, Zdanski C, Ardeshirpour F, Patel M, Hart CF, Drake AF. Recurrent infantile myofibromatosis: a report of conservative management and discussion of treatment strategies. Ear Nose Throat J 2008;87:E4.